



HEXA gene

hexosaminidase subunit alpha

Normal Function

The *HEXA* gene provides instructions for making one part (subunit) of an enzyme called beta-hexosaminidase A. Specifically, the protein produced from the *HEXA* gene forms the alpha subunit of this enzyme. One alpha subunit joins with one beta subunit (produced from the *HEXB* gene) to form a functioning enzyme.

Beta-hexosaminidase A plays a critical role in the brain and spinal cord (central nervous system). This enzyme is found in lysosomes, which are structures in cells that break down toxic substances and act as recycling centers. Within lysosomes, beta-hexosaminidase A forms part of a complex that breaks down a fatty substance called GM2 ganglioside.

Health Conditions Related to Genetic Changes

Tay-Sachs disease

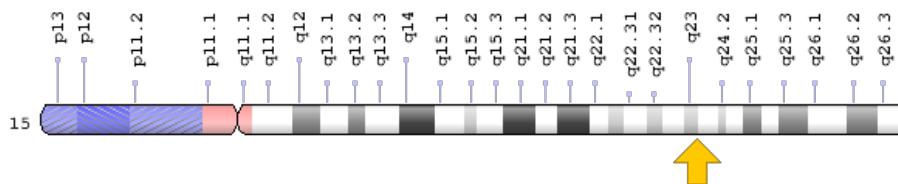
More than 120 mutations that cause Tay-Sachs disease have been identified in the *HEXA* gene. These mutations reduce or eliminate the activity of the enzyme beta-hexosaminidase A, which prevents the enzyme from breaking down GM2 ganglioside. As a result, this substance builds up to toxic levels, particularly in nerve cells in the brain and spinal cord. Progressive damage caused by the buildup of GM2 ganglioside leads to the destruction of these cells, which causes the signs and symptoms of Tay-Sachs disease.

Most of the known *HEXA* mutations result in a completely nonfunctional version of beta-hexosaminidase A. These mutations cause the severe form of Tay-Sachs disease, which appears in infancy. Other mutations reduce but do not eliminate the activity of beta-hexosaminidase A; these genetic changes are responsible for the less severe forms of Tay-Sachs disease, which appear later in life.

Chromosomal Location

Cytogenetic Location: 15q23, which is the long (q) arm of chromosome 15 at position 23

Molecular Location: base pairs 72,343,435 to 72,376,473 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Beta-hexosaminidase A
- beta-N-Acetylhexosaminidase A
- Hex A
- HEXA_HUMAN
- hexosaminidase A (alpha polypeptide)
- N-acetyl-beta-glucosaminidase

Additional Information & Resources

GeneReviews

- Hexosaminidase A Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK1218>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HEXA+AND+hexosaminidas e%5BTIAB%5D%29+OR+%28hexosaminidase+A%5BTIAB%5D%29%29+OR+ %28%28Beta-hexosaminidase+A%5BTIAB%5D%29+OR+%28beta-N-Acetylhexo saminidase+A%5BTIAB%5D%29+OR+%28N-acetyl-beta-glucosaminidase %5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND +%22last+1800+days%22%5Bdp%5D>

OMIM

- HEXOSAMINIDASE A
<http://omim.org/entry/606869>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_HEXA.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=HEXA%5Bgene%5D>
- HEXdb: Mutations in the HEXA gene
<http://www.hexdb.mcgill.ca/?Topic=HEXAdb>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4878
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3073>
- UniProt
<http://www.uniprot.org/uniprot/P06865>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/HEXA>

Reviewed: September 2008

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services